Medline

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Plasmolipin: genomic structure, chromosomal localization, protein expression pattern, and putative association with Bardet-Biedl syndrome.

Hamacher M, Pippirs U, Köhler A, Müller HW, Bosse F
Mamm Genome 2001 Dec 12:933-7

BROWSE : Mamm Genome • Volume 12 • Issue 12
VIEW : **MEDLINE**, full MEDLINE, related records

Abstract

Plasmolipin is a membrane protein and belongs to the tetraspan molecule (4TM) family, an expanding group of myelin proteins many of which could be linked to human hereditary demyelinating neuropathies. We have cloned and sequenced the mouse plasmolipin gene, revealing the common organization of the 4TM gene group with four exons and a large first intron. Western blot analysis with an antibody raised against the C-terminal intracellular part of the protein showed that plasmolipin is expressed not only in the nervous system and kidney, but also in a number of other tissues such as thymus, testis, lung, and thyroid gland. By means of radiation hybrid mapping and FISH analysis, we could localize the human plasmolipin gene to Chromosome 16q13 within the putative region of the Bardet-Biedl syndrome type 2 (BBS2) gene locus. BBS2 is a clinically and genetically heterogeneous group of disorders resulting in rod-cone dystrophy, obesity, postaxial polydactyly, renal dysfunction, and mental retardation, which were very recently associated with a novel gene designated BBS2. With respect to intrafamiliar variations in the manifestation of BBS, we suggest that plasmolipin might be either another candidate gene or a modifier of the BBS2 phenotype.

MeSH

Animal; Bardet-Biedl Syndrome; Chromosome Mapping; Chromosomes, Human, Pair 16; Comparative Study; Gene Expression Regulation; Genes, Structural; Hamsters; Human; In Situ Hybridization, Fluorescence; Mesocricetus; Mice; Multigene Family; Organ Specificity; Proteolipids; Radiation Hybrid Mapping; Rats; Species Specificity; Support, Non-U.S. Gov't

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PATENT COOPERATION TREATY

PCT

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INTERNATIONAL SEARCH REPORT

(PCT Article 18 and Rules 43 and 44)

Applicant's or agent's file reference GALO011/01WO	FOR FURTHER ACTION	see Notification of Transmittal of International Search Report (Form PCT/ISA/220) as well as, where applicable, item 5 below.
International application No. PCT/US01/17812	International filing date (*day/month/year*) 01 June 2001 (01.06.2001)	(Earliest) Priority Date (*day/month/year*) 02 June 2000 (02.06.2000)
Applicant MCINTIRE, et al		

This international search report has been prepared by this International Searching Authority and is transmitted to the applicant according to Article 18. A copy is being transmitted to the International Bureau.

This international search report consists of a total of 4 sheets.

☒ It is also accompanied by a copy of each prior art document cited in this report.

1. **Basis of the Report**

a. With regard to the **language**, the international search was carried out on the basis of the international application in the language in which it was filed, unless otherwise indicated under this item.

☐ the international search was carried out on the basis of a translation of the international application furnished to this Authority (Rule 23.1(b)).

b. With regard to any **nucleotide and/or amino acid sequence** disclosed in the international application, the international search was carried out on the basis of the sequence listing:

☐ contained in the international application in written form.

☐ filed together with the international application in computer readable form.

☐ furnished subsequently to this Authority in written form.

☐ furnished subsequently to this Authority in computer readable form.

☐ the statement that the subsequently furnished written sequence listing does not go beyond the disclosure in the international application as filed has been furnished.

☐ the statement that the information recorded in computer readable form is identical to the written sequence listing has been furnished.

2. ☐ **Certain claims were found unsearchable** (See Box I).

3. ☐ **Unity of invention is lacking** (See Box II).

4. With regard to the **title**,

☒ the text is approved as submitted by the applicant.

☐ the text has been established by this Authority to read as follows:

5. With regard to the **abstract**,

☒ the text is approved as submitted by the applicant.

☐ the text has been established, according to Rule 38.2(b), by this Authority as it appears in Box III. The applicant may, within one month from the date of mailing of this international search report, submit comments to this Authority.

6. The figure of the **drawings** to be published with the abstract is Figure No.

☐ as suggested by the applicant.

☐ because the applicant failed to suggest a figure.

☐ because this figure better characterizes the invention.

☒ None of the figures